Genetic Resources for Families

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This resource is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling $600,000 with 0 percent financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov.
Who is MSRGN?
Mountain States Regional Genetics Network
MSRGN’s mission is to ensure that individuals with genetic disorders and their families have access to quality care and appropriate genetic expertise and information through facilitating a professional network of:

- genetics clinics,
- key primary care practices,
- consumer advocates,
- and state health department resources.
MSRGN and WSRGN are 2 of the 7 Regional Genetics Networks

Regional Genetics Networks

Map showing various regions such as Heartland, Midwest, SERN, and others, with individuals Sylvia Mann and Kristi Wees marked.
NCC and Public Health Genetics Week

Public health genetics applies genetic and genomic information to improve public health and prevent disease.

- “What is Public Health Genetics?” May 26
- “Who is Involved in Public Health Genetics” May 27
- “Public Health Screening” May 28
- “Public Health Genetics Resources” May 29
- TWITTER CHAT- May 29th 2pm ET
MSRGN’S Structure and Members

- State Teams - 8 of them
- Genetic Ambassadors
- Advisory Committee
- Management Team

- Genetic Professionals (Geneticist and GC)
- Consumers, Families, Patients
- Individuals with a Genetic Condition, Family Leaders
- Primary Care Physicians
- Public Health Professionals

MSRGN’s “Customers” or Members
What is a Genetics Pop-Up?

Resources, educational materials and information about genetic services for individuals, families and their communities
A Gene in a Box

When we have an in-person Genetics Pop-Up this box of resources is mailed to the Genetic Ambassador who is hosting the Pop-Up!

For today we will be sharing resources ELECTRONICALLY!
Gene-In-A-Box

What is contained in the Gene-In-A-Box resources?
- Gene (DNA) kit with Figurine (Link to purchase)
- MSRSN magnets: English and Spanish
- MSRSN brochures: English and Spanish
- Developmental Delay Algorithm and Genetics Resource
  - Approach to Genetics Diagnosis in Developmental Delay
  - Approach to Genetics Diagnosis in Intellectual Disability
  - Non-syndromic Causes of Developmental Delay and Disability
  - Laboratory Information Genetics Labs that Perform Pre-Authorization
  - Elements of Consent for Genetic Testing
  - Laws Pertaining to Genetic Information
- Bullet point list of genetic discrimination laws
Top 10 Genetic Resources

To access electronic versions of the resources we will be sharing: scan this code or use your camera on your smart phone and focus it on this code to open the website.
TeleWhat?

Infographic from National Genetic Education Support Center
Partnership with Genetic Alliance, Family Voices, & Expecting Health
Webinar:
https://www.youtube.com/watch?v=eYHfPq_O3X4&feature=youtu.be
Top Genetic Resource:
GARD Information Center and Hotline: Genetic and Rare Disease
GARD Hotline

Available in English and Spanish
Available Monday - Friday, 12 noon-6 p.m. Eastern Time
Speak with a Genetic Counselor about a Genetic Condition
No Medical Advice can be given

1-888-205-2311
Top Genetic Resource:

National Geographic: Your Genes- 100 things you never knew!
Top Genetic Resource:
DNA Double Helix model Kit & Giant Microbes Stuffed DNA
Available for $10-15 each, online
Top Genetic Resource:
From our Colleague's at Midwest Genetics Network
Introduction to Healthcare Through Telemedicine parts 1-3
Also includes a Family Video about their Telegenetics experience
Top Genetic Resource:

NIH Genetics Curriculms:
Rare Diseases and Scientific Inquiry Grades 6-8
Human Genetic Variation Grades 9-12
Top Genetic Resource:

Genetic Alliance Books:
A Guide to Family Health History
A Guide to Genetics and Health
#4: Genetic Alliance Booklets

A Guide to Genetics and Health

A Guide to Family Health History

Available in both English and Spanish
Top Genetic Resource:

Documentary:

KEN BURNS PRESENTS THE GENE: AN INTIMATE HISTORY

part 1 & 2 free to view on PBS
Aired in April 2020
On PBS
2-part series
Produced by award winning film maker: Ken Burns
Available for FREE to watch at this website:
www.pbs.org/kenburns/the-gene/watch
Top Genetic Resource:

CDC’s Learn the Signs Act Early Milestone Tracker App
Top Genetic Resource:

MSRGN’s Developmental Delay Algorithm Toolkit
#1 Developmental Delay Algorithm Toolkit

- One of the highest referral categories to Genetics in our region is for Developmental Delay diagnosis.

- This algorithm was created to help Pediatricians work with families on beginning the genetics workup for a child with a developmental delay.

- Online Toolkit includes info for primary care doctors and families.

Synopsis of Approach to Genetic Diagnosis in Developmental Delay:

- Primary care providers may consider initiating genetic diagnostic testing for patients with developmental delay when access to genetic services is limited.
- The American Academy of Pediatrics has published their statement on appropriate genetic evaluation of children with developmental delay; see Moeensch et al Pediatrics 2014.
- Ensure that acute neurological concerns are addressed with appropriate evaluation and not deferred until after genetic testing.
- Referrals for developmental therapies should be made concurrently with a genetic evaluation.

Our recommendations are as follows:

- **Tier 1**
  - Chromosomal microarray
  - Fragile X trinucleotide repeat analysis

- **Tier 2**
  - Large gene sequencing panel of developmental delay-associated genes

- **Tier 3**
  - Whole exome or whole genome sequencing may be considered.
  - See www.treatable-is.org for a testing algorithm and information on diagnoses that have specific treatment and can be diagnosed with biochemical testing.
  - Serum amino acids
  - Serum homocysteine
  - Urine creatine metabolites
  - Urine organic acids
  - Urine purines and pyrimidines
  - Urine oligosaccharides
  - Urine mucopolysaccharides

Genes: What They Are and What They Do:

Each cell in the human body contains about 25,000 to 35,000 genes. They are found on tiny spaghetti-like structures called chromosomes, which are found inside cells. Your body is made of billions of cells, which are the very small units that make up all living things.

Most cells have one nucleus. The nucleus is a small egg-shaped structure inside the cell and it has many parts. The nucleus acts like the brain of the cell. It tells every part of the cell what to do. It contains our chromosomes and genes. As tiny as it is, the nucleus has more information in it than the biggest dictionary you’ve ever seen.

Chromosomes come in matching pairs and there are hundreds — sometimes thousands — of genes in just one chromosome. In humans, each cell nucleus contains 46 individual chromosomes or 23 pairs of chromosomes. Half of these chromosomes come from one parent and half come from the other parent.

Under the microscope, we can see that chromosomes come in different lengths and striping patterns. When they are lined up by size and similar striping pattern, the first twenty-two of the pairs are called autosomes; the final pair of chromosomes is called sex chromosomes, X and Y. The sex chromosomes determine whether you’re a boy or a girl. Females have two X chromosomes while males have one X and one Y.
Top Genetic Resource:

MSRGN’s COVID-19 Resource Sheets for families impacted by genetic conditions in our 8 states.
**Colorado Resources**

**Covid 19**

**Colorado Resources**

**Colorado Resources**

For Those Impacted by Genetic Conditions

**Know the Facts**

To find the latest COVID-19 facts, statistics and guidance from a.
- ColoradoMed
- Colorado Pediatric Specialist
- ISAM Federal
- World perspective

Please visit these websites listed below.

**Emergency Plans**

The best time to plan for an emergency is before it happens. Take time now with your loved ones to prepare an emergency plan for their specific health needs.
- Emergency Information Form (AFSP)
- Emergency Presentation for those with Metabolic Disorders
- Emergency Cards for Genetic and Metabolic Conditions

**Condition Specific**

If you or your loved one has been diagnosed with a specific genetic condition please reach out to your healthcare provider or to the patient support organization for that condition for specific information or advice.

The website www.NGC.org offers resources and support for patients and families affected by a genetic condition.

The NGSF website has some other useful links for those with genetic conditions.

**Food, Meds & Insurance**

Having an extra 30-60 days supply of food, medication, medical or food or insurance is advisable for any emergency situation.

- Unemployment
- Federal Nutrition
- Food Access: Specialized Com
- Medical Foods Access
- Medication Access:0800 Temporary

Insurance Claims for one-time early claims.

**Support**

Having a support system in place can really enhance mental health during times of crisis.

Below are some organizations that are available for support in times of need.

- Colorado Crisis Services 1-888-258-2973
- Text the word "TEXT" to 888258

For more information, please visit the websites below.

**TeleHealth**

Many primary care clinics and specialty clinics are offering telehealth visits.

- Texas Parent to Parent
- Texas Family Network (TANAP)
- Northwest LVH Texas

For more information, please visit the websites below.

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If you have any questions or would like to get involved in our activities, please contact us:

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